Elizabeth Carter, MD
Executive Director
Virginia Department of Health Professions

Dear Dr. Carter:

The Virginia Association of Genetic Counselors (VaAGC) respectfully requests that the Virginia Department of Health Professions initiate a study that assesses the need for licensure of genetic counselors in the state of Virginia.

The VaAGC believes that licensure is the only level of regulation that would provide adequate protection for the public. Genetic issues are complicated, and the field of medical genetics is rapidly changing. There is potential for harm when incorrect or incomplete genetic information is provided to patients and applied to medical decision-making. The VaAGC believes that licensure will ensure that the public, medical providers and health plans will have a mechanism to identify and access quality services from qualified genetic counselors. Such genetic counselors will have obtained appropriate training and continuing education regarding medical genetics and genetic testing. The VaAGC also believes that licensing genetic counselors will attract more high quality individuals into the profession. This will help Virginia meet the growing need for medical practitioners who are adequately trained to provide genetic consultation services.

Please refer to the attached documents that we believe provide support for our claim that genetic counselors meet the Virginia Department of Health Professions' criteria for regulation by licensure.

Sincerely

Tahnee Causey, MS, CGC
President, VaAGC

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ACRONYMS USED IN THIS PROPOSAL

ABGC	American Board of Genetic Cousnelors
ACMG	American College of Medical Geneticists
EVMS	Eastern Virginia Medical School
GIVF	Genetics and IVF Institute
LPPC	Licensure and Public Policy Committee (a VaAGC Committee)
NSGC	National Society of Genetic Counselors
VaAGC	Virginia Association of Genetic Counselors
VCU	Virginia Commonwealth University
VDHP	Virginia Department of Health Professions
UVA	University of Virginia—Charlottesville

CONTACT INFORMATION FOR PROPOSAL SUBMITTER

On behalf of the VaAGC, I respectfully submit this request.

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SUMMARY OF PROPOSAL PREPARATION

This proposal was co-written by members of the Virginia Association of Genetic Counselors (VaAGC) Licensure and Public Policy Committee and the National Society of Genetic Counselors (NSGC). This proposal has been approved by the VaAGC members at large.

The VaAGC was formed in May of 2008. General information about the VaAGC can be found at www.vaagc.org. There are approximately 60 genetic counselors who live or work in the Commonwealth of Virginia. All of these genetic counselors were invited to become members of the VaAGC. There is currently one genetic counselor in the state who wished to opt out of membership. The general VaAGC membership elected seven officers to represent and promote the interests of genetic counselors in our state. Both elected and unelected member volunteers serve on various committees that have been established by the officers. Due to the small size of the group, committees elect their own chairs. The Licensure and Public Policy Committee (LPPC) is made up the following VaAGC members:

- Jen English, LPPC Chair
- Tahnee Causey, VaAGC officer, President
- Kara Bui, VaAGC officer, Past President and LPPC Co-Chair
- Christine Seward, VaAGC officer, Northern Virginia Representative
- Selvi Sriranganathan, VaAGC member
- Kara Withrow, VaAGC member
- Karen Ventura, VaAGC member
- Asheley Supik, VaAGC member

The LPPC was established in part to research the feasibility of licensure for genetic counselors in Virginia. The LPPC recommended that the VaAGC submit a proposal to the Virginia Department of Health Professions to request a study assessing the need for licensure of genetic counselors. This recommendation was based on surveys given to the Commonwealth's genetic counselors in 2006 and 2007, which showed majority support for regulation. The LPPC drafted this proposal, which was then approved by the VaAGC officers and the National Society of Genetic Counselors (NSGC). The VaAGC officers then provided the proposal to the general VaAGC membership and requested written comments regarding support for or against the formal regulation of genetic counselors in this state. Two months were provided for the membership to send the VaAGC officers comments. Suggestions for changes were received from five genetic counselors. All of the suggestions were incorporated into this document where applicable. The VaAGC officers actively sought out members who disapprove of submitting this proposal to the VDHP. No members who disapprove were found. Members who approve of the submission of this document initialed printed copies of the proposal. On March 18, 2010, the VaAGC officers unanimously voted to submit the following proposal to the Virginia Department of Health Professions.

PROFILE OF GENETIC COUNSELORS IN VIRGINIA

What is a Genetic Counselor?

A genetic counselor is a health care professional who has specific academic training to serve and support individuals and families with genetic conditions or those who may be at risk for genetic conditions or birth defects. Genetic counselors typically practice as part of a health care team, primarily in prenatal, pediatric and oncology settings. Many genetic counselors work in industry, such as laboratories that offer genetic testing services. Other genetic counselors work in public health settings educating the public about screening and prevention options for certain birth defects and genetic conditions. The VaAGC agrees with the American Board of Genetic Counselors' (ABGC) description of a genetic counselor:

> *"[Genetic counselors] interpret and provide clear and comprehensive information about the risk of any medical condition that may have a genetic contribution. They ascertain the usefulness of genetic technologies for individual families and facilitate an informed decision-making process that elicits and respects the spectrum of personal beliefs and values that exist in society."*

General Consumer Information About Genetic Counseling

Making Sense of your Genes: A Guide to Genetic Counseling
www.nsgc.org/client_files/GuidetoGeneticCounseling.pdf

Genetic Counseling (Prenatal/Pediatric Focus)
www.marchofdimes.com/pnhec/4439_15008.asp

Can You Benefit From Genetic Counseling? (Prenatal/Pediatric Focus)
www.marchofdimes.com/pnhec/4439_1088.asp

Cancer Genetic Counselors
www.nsgc.org/members_only/sig/cancersigbroch2.pdf

What is a Genetic Counselor's Scope of Practice?

The VaAGC agrees with the NSGC's complete definition of a genetic counselor's scope of practice, which can be found at www.nsgc.org/client_files/SOP_final_0607.pdf. An excerpt of this document follows:

> *"Genetic counselors are health professionals with specialized education, training and experience in medical genetics and counseling who help people understand and adapt to the implications of genetic contributions to disease. Genetic counselors interact with clients and other healthcare professionals in a variety of*

clinical and non-clinical settings, including but not limited to university-based medical centers, private hospitals, private practice, and industry settings. The instruction in clinical genetics, counseling, and communication skills required to carry out the professional responsibilities described in this statement is provided in graduate training programs accredited by the American Board of Genetic Counseling (ABGC) or the equivalent, as well as through professional experience and continuing education courses. The responsibilities of a genetic counselor are threefold: (i) to provide expertise in clinical genetics; (ii) to counsel and communicate with patients on matters of clinical genetics; and (iii) to provide genetic counseling services in accordance with professional ethics and values."

Genetic Counselors in Virginia

There are approximately 60 genetic counselors living and/or working in Virginia. More than 80% of the genetic counselors work at least part-time in a clinical setting and see patients as part of a multidisciplinary medical team. Clinical genetic counselors identify individuals who are at risk for genetic conditions, analyze inheritance patterns, calculate the potential for recurrence of a given genetic condition and educate families about medical management options based on the risk assessment. Clinical genetic counselors may also collaborate with the medical team to diagnose genetic conditions—particularly when a genetic test is recommended and the test results indicate that the individual is or is not a carrier for that condition. Less than 20% of genetic counselors in Virginia work at least part-time in non-clinical settings providing genetic services but not routinely participating in direct patient care.

CRITERION ONE: RISK FOR HARM TO THE CONSUMER

The VaAGC believes that licensing genetic counselors will prevent harm to the consumer by:

- increasing the public's awareness of what constitutes quality genetic counseling services and who is appropriately trained to provide these services
- improving the public's access to quality genetic counseling services
- allowing the public to have a method of recourse for unprofessional genetic counseling services without relying upon malpractice litigation, which is costly and time consuming.

There are currently over 1,200 genetic tests for human genetic conditions available through commercial laboratories and in research settings. Some of these tests are used to diagnose conditions. Others are used to help currently healthy individuals know if they have an increased risk for a particular genetic condition. A wide variety of technologies are used to extract information from DNA and chromosomes. It is undeniably difficult for general health practitioners to stay current on the availability of new genetic tests, let alone the benefits, limitations and adverse effects related to each test. Previous studies have shown that primary care providers and specialists lack both a fundamental knowledge of genetics as well as continuing educational opportunities in the field. This same issue is reflected in the allied health professions, where studies have found that over 75% of individuals rated their training in genetics as marginal or none; however over 40% still discussed genetic inheritance, testing and genetic conditions. [Christianson CA et al., 2005; Lapham EV et al., 2000]

It is helpful for general health practitioners to refer their patients to an appropriately trained genetic counselor, who can help the provider know 1) when a family history shows evidence of a hereditary condition; 2) when patient is an eligible candidate for genetic testing or medical interventions based on family history interpretation; 3) how to select the best genetic test for that patient; and 4) how to recognize the limitations of the test information when developing a medical management plan for that patient. In addition to supporting the general health practitioner, a genetic counselor is trained to help individuals and families make informed decisions about genetic testing. Major medical associations and health plans recognize the importance of pre- and post-test counseling for individuals and families who undergo genetic testing. (See Table 1 on next page.) Appendix Two contains an annotated bibliography of statements regarding genetic counseling from various professional medical societies.

The sequencing of the human genome has led to an explosion of new genetic tests and a corresponding increase in demand from the public for these tests. Despite the increased demand, there has not been a corresponding increase in access to providers who are sufficiently trained to provide genetic services. Other states that license genetic counselors found that the number of genetic counselors practicing in the state rose following licensure. For example, Utah began licensing genetic counselors in 2002.

Prior to 2002, there were approximately 14 genetic counselors in the state. After licensure, the number of genetic counselors doubled to 30.

Table 1. Policy Statements from Major Medical Associations and Health Insurers

- **Aetna Clinical Policy Bulletin: Genetic Counseling**
  http://www.aetna.com/cpb/medical/data/100_199/0189.html

- **American Medical Association (AMA) statement on genetic testing**
  http://www.ama-assn.org/ama/pub/physician-resources/medical-science/genetics-molecular-medicine/related-policy-topics/genetic-testing.shtml

- **American Society of Clinical Oncology policy statement update: genetic testing for cancer susceptibility.**
  J Clin Oncol. 2003 Jun 15;21(12):2397-406. Epub 2003 Apr 11.
  http://jco.ascopubs.org/cgi/content/full/21/12/2397

  "ASCO supports efforts to ensure that all individuals at significantly increased risk of hereditary cancer have access to appropriate genetic counseling…"

- **Cigna Medical Coverage Policy: Genetic Counseling**
  http://www.cigna.com/customer_care/healthcare_professional/coverage_positions/medical/mm_0297_coveragepositioncriteria_genetic_counseling.pdf

- **United Healthcare News Bulletin**
  http://content.4at5.net/email_domains/unr/47655/hosted/clinician_2.html

  UHC implements a new policy ensure access to genetic counselors for patients seeking BRCA1 and BRCA2 genetic testing in part because *"… many genetic tests are ordered and performed with incomplete information, unclear indications, and insufficient support services such as test interpretation and genetic counseling."*

What harm can happen when an individual does not receive quality genetic counseling services?

- Misinterpretation of pedigree and/or genetic test results which can lead to
  - unnecessary medical treatment and/or surgery
  - lack of prevention disease monitoring strategies
  - irreversible reproductive decisions
- Misinterpretation of genetic risks (or lack of risks)
- Emotional harm related to insufficient pre- or post-test counseling
  - avoidable fear, anxiety, guilt
- Unnecessary genetic testing, which can be costly

Harm can be subcategorized as economic harm, medical harm, psychological harm, harm due to lack of awareness and/or harm due to lack of recourse.

ECONOMIC HARM: Inappropriate and unnecessary genetic testing contributes to increasing societal health care costs in general. Individual consumers can be held personally responsible for the costs of genetic testing that are not medically indicated.

Provision of costly health interventions based upon inaccurate interpretation of genetic information can have dire economic effects. For example, we are aware of a SW Virginia family whose insurance was billed over $3,000 for genetic testing, when the most appropriate test actually cost no more than $450. Recently, genetic testing for cystic fibrosis carrier status has been initiated on a widespread basis for all couples currently pregnant or anticipating a pregnancy. It has been documented that non-genetic medical practitioners have incorrectly interpreted these test results, leading to unnecessary prenatal test procedures (amniocentesis) which are costly and infer a risk for pregnancy loss [Redman JB et al., 2003]. Licensing of genetic counselors would reduce costs, as their clinical expertise allows them to critically evaluate the appropriateness and utility of genetic tests.

There is a growing trend for genetic tests to be offered directly to consumers online. Some of the tests are considered non-medical. For instance, there are genetic tests used to determine paternity or ancestry. Other genetic tests, however, are specifically marketed to the public to predict health risks or to diagnose medical conditions. There is a potential for medical harm from direct-to-consumer genetic tests that aim to provide health information for consumers when there is insufficient medical oversight by health care providers with adequate training in genetics. There is also potential for economic harm when consumers purchase tests based on misleading/dubious claims or when they make medical management decisions based on misinterpreted results or results from clinically invalid tests. The American Society of Human Genetics and the American College of Medical Genetics have published statements of direct-to-consumer genetic testing in the United States [ASHG, 2007 and ACMG]. Both groups promote the use of safeguards to help ensure that consumers using these types of tests have appropriate informed consent and that there is federal oversight of the clinical and analytic validity of such genetic tests.

MEDICAL HARM: Inaccurate or inappropriate medical care could potentially harm any of the patients with whom an inadequately trained health professional has contact. One example of incorrect counseling and test interpretation is found in the *New England Journal of Medicine* [Giardiello FM et al., 1997]. This article highlights the likelihood of harm when non-genetic physicians misinterpret genetic testing information. The authors of this study surveyed 177 patients who underwent genetic predisposition testing for Familial Adenomatous Polyposis (FAP), an inherited condition that leads to the development of colon cancer early in life. This cancer can be prevented with appropriate surgical intervention, so accurate interpretation of test results is crucial. Eighteen percent of the patients studied underwent genetic counseling prior to genetic testing and these patients received accurate interpretation of their genetic test results. Of the remaining 145 patients who did not receive genetic counseling, 30% were given the wrong test interpretation because caregivers incorrectly interpreted inconclusive test results as meaning that the patient definitely did not have an FAP mutation. The consequences of this misinterpretation are potentially devastating, since these individuals would have thought they were no longer at risk for colon cancer and were likely to stop potentially life-saving endoscopic screening.

Harm can also occur if practitioners do not elicit complete family history information. This was demonstrated in a study in the *Journal of Perinatology*, which assessed the adequacy of genetic risk assessment among primary care providers [Cohn GM et al., 1996]. This study found that in 35% of the 378 cases studied, significant genetic risk that had been missed by the referring physician was identified in a subsequent genetic consultation. Failure to identify significant genetic risks may lead to inappropriate medical management, physical injury, death or psychological distress. The concern for harm has already been recognized by the general medical-legal community as evidenced by the cost of malpractice suits against non-genetic physicians for failure to inform patients of heritable risks to themselves or their offspring [Pelias MZ, 1992 and Deftos LJ, 1998].

PSYCHOLOGICAL HARM: Individuals affected by genetic conditions often face serious social and psychological challenges. Genetic testing is unique in that it does not just impact the individual, it often affects the whole family. At minimum, parents may feel guilty or stigmatized when they pass on non-working genes to their children. If testing is done without appropriate preparation and informed consent, the risks may outweigh any potential benefits. Individuals undergoing pre-symptomatic genetic testing, may experience overwhelming anxiety regarding their at-risk status and the impending threat from the disease process itself. Risk assessment and counseling and testing should be performed in a timely, appropriate, family-centered and culturally competent manner, while at the same time accurately communicating genetic risk. Previous studies have shown even with simple case histories, non-genetic health care providers routinely assess a patient's risk inaccurately, often overestimating an individual's risk [Fry A et al., 1999]. Overestimation of genetic risk can result in significant psychological consequences [Butow et al., 2003]. Genetic counselors receive unique training permitting them to anticipate and mitigate psychological harms associated with genetic conditions and genetic testing. Genetic counselors are the most knowledgeable about genetic developments and already utilize family systems methodologies; therefore, licensing genetic counselors would be the most effective way of minimizing psychological harm to the public.

LACK OF AWARENESS HARM: The aforementioned cases demonstrate that the majority of consumers do not possess the comprehensive knowledge or experience necessary to evaluate whether a genetic counselor is competent. The extent of the public's knowledge of genetics is limited and often skewed by media resources. A survey of key stakeholders in Virginia found that over 75% of those surveyed rated the genetic knowledge level of the average Virginian to be poor or very poor [Bodurtha JN et al. 2006]. Given the rapid pace of genetic discoveries using advanced DNA technology, it is difficult for an individual not trained in genetics to understand information about their unique condition. Information about any one condition is rapidly changing and highly specialized, requiring ongoing education to be able to interpret new tests and data. In addition, many potential genetic counseling clients are not aware of the availability or the scope of this service. State regulation of the profession will help increase public awareness of genetic counseling services. State regulation will also provide potential consumers with assurances of competency.

LACK OF RECOURSE HARM: Licensure holds genetic counselors accountable for their actions and allows for legal recourse if inappropriate care is provided. With licensure, the Commonwealth of Virginia will legally recognize a genetic counselor's specific scope of practice and the standards for professional conduct. If a licensed genetic counselor violates the laws or rules defined by the legislation, then Virginia has the authority to take disciplinary action. Currently, there are no existing laws in Virginia or at the federal level that provide the public with a mechanism to report incompetent, unethical, unlawful behavior of a genetic counselor or to sanction a genetic counselor for proven offenses of these claims and/or for operating outside of their scope of practice.

CRITERION TWO: SPECIALIZED SKILLS AND TRAINING

Genetic counselors have specialized graduate-level training in genetic counseling. Most genetic counselors have a Master of Science degree in genetic counseling. In the US, the Master of Science in Genetic Counseling is a terminal degree program for individuals providing clinical genetic counseling services. Virginia has one accredited genetic counselor training program at Virginia Commonwealth University (VCU) in Richmond. There are 30 other accredited genetic counseling programs in the US. In order to be an accredited genetic counseling program, the American Board of Genetic Counselors (ABGC) requires that diplomates demonstrate specific competencies that should have been developed through graduate coursework in:

- human, medical and clinical genetics
- psychosocial theory and techniques
- social, ethical and legal issues
- health-care delivery systems and public health principles
- teaching techniques
- research methods
- clinical training, working with individuals and families affected with a broad range of genetic conditions.

Genetic counselors receive training in legal, ethical and social principles pertaining to the delivery of genetic services, as well as training in professional ethics and values. Genetic counselors adhere to a code of ethics specifying that the counselor-client relationship be based on values of care and respect for the client's autonomy, individuality, welfare and freedom. The primary concern of genetic counselors is the interests of their clients. The Code of Ethics of the National Society of Genetic Counselors states that genetic counselors strive to:

- equally serve all who seek their services
- respect their clients' beliefs, cultural traditions, inclinations, circumstances and feelings
- enable their clients to make informed independent decisions, free of coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences
- refer clients to other competent professionals when they are unable to support their clients, maintain as confidential any information received from clients, unless released by the client
- avoid the exploitation of their clients for personal advantage, profit or interest.

In addition to accrediting genetic counseling graduate programs, the ABGC offers a private, non-governmental credential for people who provide services in the medical genetics specialty of genetic counseling. Since 1992, the ABGC has administered a voluntary certification exam for diplomates (prior to this, the American Board of Medical Genetics (ABMG) alone certified genetic counselors). ABGC develops the examination

and offers it annually. New graduates are eligible to apply for active candidate status to take the certification exam for up to three exam cycles within five years of graduation. The certification process includes verification of specialized training and documentation of clinical experiences under the supervision of an ABGC-certified genetic counselor or ABMG certified clinical geneticist. Evaluation and counseling of 50 different patients and/or families is required. The 50 cases must include three areas: fetal risk assessment, diagnostic evaluation management and genetic risk assessment. Appropriate supervision of students is required. Of the approximately 60 genetic counselors practicing in Virginia, there are at least two who do not have ABGC certification and are not eligible to sit for a future exam.

While individual genetic counselors may choose to specialize in particular areas of genetic medicine, such as prenatal genetics or hereditary cancer, the ABGC does not offer subspecialty certifications. Also, there are no accredited training programs for genetic counseling subspecialities. The VaAGC believes that "generic" regulation of all genetic counselors regardless of subspeciality is the most practical solution for preventing harm to the consumer, as core components of quality genetic counseling are consistent across areas of specialty.

An unqualified provider's lack of awareness of genetic advances may deprive patients of valuable information and medical options. For example, one study found that 57% of clinicians did not recognize that a patient with multiple relatives affected with breast cancer on her father's side should be considered at increased risk for breast and ovarian cancer [Hayflick SJ et al., 1998]. In a specific case (*Seattle Times*, October 19, 2006) a woman with a strong family history of breast cancer and a personal history of breast cancer at age 28 and 37 subsequently died of ovarian cancer. Virginia Mason Medical Center settled the case after medical experts testified that the patient clearly had a hereditary breast and ovarian cancer syndrome and should have been offered the option of removing her ovaries, in an effort to prevent her third cancer and subsequent death. Furthermore, most medical professionals have little training in medical genetics and are familiar with only the most common genetic disorders. A survey of US Medical schools revealed that the average medical student was exposed to only 29 hours of didactic coursework in medical genetics [APHMG, 1998]. While nearly 70% of allied health professionals surveyed reported discussing genetic issues with clients, 80% had no formal genetics training [Lapham et Al., 2000]. As a result, few medical professionals understand the natural history of most genetic conditions, the complexity of genetic tests or the interpretation and implications of test results. Clinical genetic knowledge is rapidly expanding into common complex disorders (such as cancer). The expansion of genetics into the care of common disorders will only make it more difficult for the busy clinician to stay informed about genetic advances relevant to their practice. While there are some licensed non-genetic health care providers who can provide aspects of genetic counseling, most of these professionals find it difficult to stay abreast of constantly evolving genetic knowledge and technology.

Often, issues of harm not only apply to the patient but also to extended family members. Genetic counselors communicate risk information regarding genetic conditions to

individuals and their relatives. This information is based on a thorough assessment of the family medical history and interpretation of genetic test results. Patients and their physicians use this genetic information to make medical decisions. Unskilled health care providers may inadequately collect family history, inaccurately assess risk and incorrectly interpret diagnostic evaluations.

A fundamental tenet of genetic counseling is the principle of non-directiveness whereby medical information is provided to a patient in a non-biased manner, allowing each individual to make autonomous medical decisions based upon his/her own belief system. Non-directiveness as a fundamental principle of the profession sets genetic counseling apart from many other health care providers. There has been well-documented research which indicates that other health care professionals are more likely to provide directive counseling (i.e., making the patient's decision for them), thereby not allowing for the client to base decisions upon their own value-based system [Geller G et al., 1993].

For example, a study in the *Journal of the American Medical Association* [Zuckerman S et al., 2007] found that couples whose fetuses were affected with Gaucher disease were far less likely to terminate their pregnancies if they consulted with a genetic counselor. The study stated that genetic counseling helps most accurately communicate the low risk of developing symptoms of this disease, the severity of the disease and the fact that it is treatable.

CRITERION THREE: THE FUNCTIONS AND RESPONSIBILITIES OF THE PRACTITIONER REQUIRE INDEPENDENT JUDGMENT AND THE MEMBERS OF THE OCCUPATIONAL GROUP PRACTICE AUTONOMOUSLY

Most genetic counselors see individuals for patient care because another health care provider has referred the individual for genetic services. It is expected that the genetic counselor will make calculations regarding the level of risk for a genetic condition with the intent to guide the patient's medical management. The genetic counselor's risk assessment may lead to the recommendation that the patient consider a particular genetic test. If the patient elects testing, then the test result may be interpreted by the genetic counselor. Certain genetic tests are the sole basis for the diagnosis of a genetic condition. Likewise, the genetic counselor's risk assessment may be the basis for a specific medical management plan that would be administered by a physician or nurse practitioner. Genetic counselors often work with vulnerable populations, such as pregnant women, individuals with disabilities and individuals with significantly compromised health.

Genetic counselors in clinical settings typically collaborate with other licensed health professionals to assist those providers and their patients with decision-making related to genetic risks. It is a licensed health care provider who would be responsible for referring, ordering, prescribing or administering a treatment plan based on the genetic counselor's evaluation of the patient.

ABGC-accredited genetic counseling programs require genetic counseling students to be supervised by ABGC certified genetic counselors.

The NSGC scope of practice for a genetic counselor does not include using dangerous equipment or substances when performing standard functions.

The institution that employs the genetic counselor is legally accountable/liable for acts performed under supervision.

CRITERION FOUR: THE SCOPE OF PRACTICE IS DISTINGUISHABLE FROM OTHER LICENSED, CERTIFIED AND REGISTERED OCCUPATIONS

Genetic counseling is a process of patient and family education within a framework of non-directive communication. The specialized training and skills of a genetic counselor allow him/her to obtain a detailed family medical history, as well as provide patient education, risk assessment, genetic testing and research information, and medical management options that would not usually be included in the genetic counseling services provided by other regulated health professionals. Another key component of genetic counseling is referral to appropriate federal/state and individual or family support resources. When other health professionals find that a patient requires highly specialized genetic services, the provider may wish to refer the patient to a genetic counselor. While genetic counselors collaborate closely with their M.D. geneticist colleagues, the scope of practice of a genetic counselor is separate. Clinical geneticists complete a detailed physical examination, review imaging and test results, establish a differential diagnosis and recommend appropriate diagnostic tests. Genetic counselors use the information provided by the clinical geneticist to educate families and connect them to appropriate resources.

Licensure is intended to protect the public from harm by unqualified providers, NOT to restrict the practice of medicine. Other licensed health care professionals may continue to practice the profession for which they are trained, including counseling of patients. Because many physicians, nurses and physician assistants do not have up-to-date training in genetics or may not have the time to provide quality genetic counseling, they may prefer to refer to a genetic counselor when those services are needed; but licensure would not require them to refer to a genetic counselor. Instead, the VaAGC expects that regulating genetic counselors will allow other providers to more easily refer to genetic counselors as needed due to improved access to quality genetic counseling services.

Examples of a genetic counselor's scope of practice in various clinical settings:

1. A nurse practitioner in an obstetrics practice learns that a patient is a cystic fibrosis carrier following routine ethnic carrier screening. The nurse practitioner refers the patient to a genetic counselor in a perinatologist's office. The genetic counselor calculates the risk that the child from the current pregnancy will have cystic fibrosis. The genetic counselor then educates the couple about cystic fibrosis, discusses prenatal and postnatal testing options and assists the couple with making an informed decision about their testing options. In this situation, the patient's husband wishes to have cystic fibrosis carrier screening before the couple makes a decision about prenatal diagnosis via genetic amniocentesis. The genetic counselor collaborates with the perinatology medical team regarding ordering the appropriate test, interpreting the test results, counseling the couple about the test results, providing informed consent and making referrals to other specialties, if needed. In this situation, the nurse practitioner provided aspects of genetic counseling services by offering cystic fibrosis carrier screening to the patient and

discussing the test results with her. The perinatologist supervised the services performed by the genetic counselor. The perinatologist's role included ordering the husband's cystic fibrosis carrier screen, and the perinatologist would perform the amniocentesis, if later elected by the patient.

2. A pediatrician refers a child to a pediatric geneticist because the child is exhibiting evidence of developmental delay and has dental abnormalities. The genetic counselor meets with the family and takes a detailed family medical history. The pediatric geneticist performs a physical exam and recommends a series of tests, including chromosome analysis. The genetic counselor assists the pediatric geneticist with arranging the necessary referrals, coordinating appropriate tests and counseling the family about the test results. The genetic counselor provides the family with local support resources related to the child's condition. In this situation, the M.D. geneticists role is focused on diagnosis and management whereas the genetic counselor's duties primarily revolve around communication of genetic information and referral to social services and family support.

3. An oncologist orders genetic testing on a patient whom he suspects may have a hereditary cancer syndrome called Lynch syndrome. The initial genetic test results are normal. The oncologist refers the patient to a genetic counselor who takes a detailed clinical and family medical history, calculates the likelihood that the patient may have Lynch syndrome due to an undetected gene mutation and calculates the likelihood that the patient may have a different hereditary cancer syndrome. The genetic counselor recommends that the oncologist order specialized tumor testing related to Lynch syndrome and assists the oncologist with ordering the appropriate test. The tumor test result is abnormal. The genetic counselor interprets the tumor test result in the context of this patient's family history. In this case, the genetic counselor believes that the patient may have Lynch syndrome despite the previous normal genetic test results. The genetic counselor recommends that the oncologist manage this patient as he would medically manage other individuals known to have Lynch syndrome. In this scenario, the oncologist provided aspects of genetic counseling when he identified that his patient was an appropriate candidate for testing, performed the informed consent for the genetic test, interpreted the results and determined that further genetic evaluation was needed. The oncologist referred to a genetic counselor because the use of genetic tests in oncology is rapidly changing and the oncologist believed that the genetic counselor's high degree of specialization would allow her to educate the patient and the oncology team about the nuances of genetic testing for Lynch syndrome.

CRITERION FIVE: THE ECONOMIC COSTS TO THE PUBLIC OF REGULATING THE OCCUPATIONAL GROUP ARE JUSTIFIED

The fees for genetic counseling in Virginia are highly variable and depend greatly upon the clinical setting. A genetic counseling CPT code 96040 became an eligible code in January of 2007. This CPT code has been reimbursed by third party payors in Virginia, but the reimbursement patterns are inconsistent. Currently, Medicare does not recognize the code as it is bundled into other Evaluation and Management codes. With licensure, the VaAGC expects that third party payor reimbursement for this CPT code will become clearer. It is not yet known if licensed genetic counselors get reimbursed for services using this CPT code at a higher rate than unlicensed genetic counselors, since the CPT code is new and genetic counselors have only been licensed in other states for a few years. The VaAGC acknowledges that third party payor recognition of the CPT code may ultimately result in higher fees for genetic counseling services, but the issues related to CPT code reimbursement will occur with or with out the regulation of genetic counselors. It is expected that reimbursement for genetic counseling services by a physician would be higher then when the same service is provided by a genetic counselor. It is not expected that reimbursement for genetic counseling services would be reimbursed at a lower rate if it were performed by another non-physician licensed health providers.

The University of Virginia (UVA), Eastern Virginia Medical School (EVMS) and Virginia Commonwealth University (VCU) are paid a flat rate each year for provision of genetic services. EVMS and UVA each use a portion of the money from this grant toward three genetic counselor salaries (six total). VCU does not use any of this particular state grant for any genetic counselor's salary. GIVF is reimbursed by the state on a procedure-by-procedure basis. Procedures are defined as amniocentesis, chorionic villus sampling, fetal ultrasound, maternal serum screening, etc. Genetic counseling is not considered a procedure and is therefore not reimbursed by the state, but there is a stipulation that GIVF, UVA, EVMS and VCU have at least two ABGC/ABMG board certified or board eligible genetic counselors on staff.

The average salary of a genetic counselor in the mid-Atlantic region of the US, which includes Virginia, is $62,238. This modest average salary means that it is important to the VaAGC that regulatory fees are within the means of our membership. It is expected that licensed genetic counselors in Virginia would be regulated by the Board of Medicine as opposed to being regulated by a separate board. If so, the VaAGC prefers that a Genetic Counseling Advisory Board be created. In states where genetic counselors are regulated by a Board of Medicine, the application fees range from $150 to $300. It is anticipated that licensure of genetic counselors in Virginia will be fiscally neutral. Given the small number of genetic counselors in Virginia, existing personnel should be able to absorb the processing of the applications and checks without hiring additional personnel. In fact, processing genetic counselor applications is anticipated to be easier for the state than other professions because the state does not have to create a genetic counseling

licensing examination. Additionally, the state would rarely have to conduct thorough reviews of the applicant if the educational background of the applicant is appropriate, as this is done by the ABGC prior to certification. Although the first year of licensing may be somewhat time and cost intensive, subsequent years are expected to provide little additional effort or expense. Utah began licensing genetic counselors in 2002. They estimate they are currently profiting from genetic counseling licensing fees.

CRITERION SIX: THERE ARE NO ALTERNATIVES TO STATE REGULATION OF THE OCCUPATION WHICH ADEQUATELY PROTECT THE PUBLIC

The VaAGC believes that licensure is the only appropriate means of regulation for genetic counselors. Currently, professional standards for genetic counselors can only be enforced by each genetic counselor's employer. This assumes that the employer is familiar enough with the standards for ABGC certification and the NSGC Code of Ethics [NSGC Code of Ethics Rev, 2006] in order to judge if their employee is adhering to those standards. It should be taken into consideration that ABGC certification is not required in order to practice as a genetic counselor in Virginia. Likewise, it is not mandatory for a genetic counselor to be a member of the NSGC, which requires its members to adhere to the NSGC Code of Ethics. At this time, there is currently no state or federal requirement for an institution in Virginia to credential a genetic counselor to ensure that he/she is able to proficiently perform his/her job.

Also, there is currently no mechanism for peer review outside of the employer and currently no way for peers to challenge a genetic counselor's competency or to effectively discipline a genetic counselor who is found to be engaging in unprofessional conduct. If a consumer is harmed by a genetic counselor's poor practice, the only avenue for recourse is malpractice litigation. Individual employers may have a list of legal offenses that may preclude a genetic counselor from being hired by that employer, but there are no legal offenses that would prevent a genetic counselor from practicing in Virginia. Title protection and registration do not protect the public from harm, as there is no offer of public legal recourse other than costly and time consuming malpractice litigation. With litigation as the only recourse of action, unqualified health care providers may not be appropriately disciplined for providing substandard care.

APPENDIX ONE: REFERENCES

ACMG Statement on Direct-to-Consumer Genetic Testing. American College of Medical Genetics. Web. Available at: http://www.acmg.net/StaticContent/StaticPages/DTC_Statement.pdf

Bodurtha JN, Teague K, Meister K et al. Virginia GeneSEAN: Genetic Services and Education-An Assessment of Needs. Final report from GeneSEAN project, funded by grant from Virginia Chapter of March of Dimes, March 2006. Available at: http://www.vahealth.org/gns/documents/2006/2006%2004%20Virginia%20GeneSEAN%20Report.pdf. Last accessed October 28, 2009.

Butow, Phyllis N. "Psychological outcomes and risk perception after genetic testing and counselling in breast cancer; a systematic review." *The Medical Journal of Australia* 178 (2003): 77-81. Web. Available at: http://www.mja.com.au/public/issues/178_02_200103/but10729_fm.pdf

Christian CA, McWalter KM and Warren NS. Assessment of Allied Health Graduates' Preparation to Integrate Genetic Knowledge and Skills into Clinical Practice. *Journal of Allied Health 2005*;34:138-144.

Cohn GM, Gould M, Miller RC et al. The importance of genetic counseling before amniocentesis. *Journal of Perinatology* 1996;16(5):352-357.

Deftos LJ. The evolving duty to disclose the presence of genetic disease to relatives. *Academic Medicine* 1998;73(9):962-968.

Fry A, Campbell H, Gudmundsdottir H et al. GPs' views on their role in cancer genetic services and current practice. *Family Practice* 1999;16(5):468-474.

Geller G, Tamor ES, Chase GA et al. Incorporation of Genetics in Primary Care Practice: Will Physicians Do the Counseling and Will They Be Directive? *Archives of Family Medicine* 1993;2:1119-1125.

Giardiello FM, Brensinger JD, Petersen GM et al. The use and interpretation of commercial APC gene testing for familial adenomatous polyposis. *New England Journal of Medicine* 1997;336:823-827.

Hayflick SJ, Eiff MP, Carpenter L et al. Primary care physicians' utilization and perceptions of genetic services. *Genetics in Medicine* 1998;1(1)13-21.

Lapham EV, Kozma C, Weiss JO et al. The gap between practice and genetics education of health professionals: HuGEM survey results. *Genetics in Medicine* 2000;2(4):226-231.

National Society of Genetic Counselors Code of Ethics. Adopted 1992, revised 2006. Available at http://www.nsgc.org/about/codeEthics.cfm. Last accessed October 29, 2009.

Pelias MZ. Duty to disclose in medical genetics: a legal perspective. *American Journal of Medical Genetics* 1991;39(3):347-354.

Redman JB, Franklin CR, LaGrave D, York M, Entwistle TD, Strom CM. Reporting the CFTR Intron 8 5T Allele in General Population Screening: Muddying the Waters. Poster Presentation, American College of Medical Genetics, 2003.

ASHG Statement on Direct-to-Consumer Genetic Testing in the United States. *The American Journal of Human Genetics* 2007(81):635-37. Available at: http://www.ashg.org/pdf/dtc_statement.pdf

Zuckerman S, Lahad A, Shmueli A et al. Carrier Screening for Gaucher Disease: Lessons for Low-Penetrance, Treatable Diseases. *Journal of American Medical Association* 2007;298(11):1281-1290.

APPENDIX TWO: STATEMENTS REGARDING GENETIC COUNSELING FROM PROFESSIONAL MEDICAL SOCIETIES

1. American Academy of Pediatrics

The American Academy of Pediatrics Committee on Children With Disabilities. (2001) The Pediatrician's Role in the Diagnosis and Management of Autistic Spectrum Disorder in Children. *Pediatrics.* 107(5): 1221-1226.
This set of guidelines, published by the AAP, is designed to assist pediatricians in recognizing and navigating their role in the diagnosis and management of autistic spectrum disorders. The AAP recommends that families should receive genetic counseling regarding recurrence risk appropriate to the etiologic diagnosis.

The American Academy of Pediatrics Committee on Genetics. (2001) Health Supervision for Children with Down Syndrome. *Pediatrics.* 107(2): 442-449.
This set of guidelines, published by the AAP, is designed to assist pediatricians in caring for children with Down syndrome. The AAP indicates that when appropriate, referral to a clinical geneticist should be considered for a more extended discussion of clinical outcomes and variability, recurrence rates, future reproductive options, and evaluation of the risks for other family members. They also recommend that if the child has a translocation and the translocation is in either parent, additional familial studies and counseling should be instituted.

The American Academy of Pediatrics Committee on Genetics. (1996) Health Supervision for Children With Fragile X Syndrome. *Pediatrics.* 98(2): 297-300.
This set of guidelines, published by the AAP, is designed to assist pediatricians in caring for children with fragile X syndrome. The AAP recommends genetic counseling and carrier detection for siblings and relatives of a child diagnosed with Fragile X syndrome.

Lee, PA, Houk, CP, Faisal, S. (2006) Consensus Statement on Management of Intrasex Disorders. *Pediatrics.* 118(2): e488-e500.
This consensus statement, published by the AAP, is designed to assist clinicians to understand the proper management of intrasex disorders. The AAP indicates that optimal care for children with intrasex disorders requires an experienced multidisciplinary team that is generally found in tertiary care centers, and includes pediatric subspecialists in endocrinology, surgery, and/or urology, psychology/psychiatry, gynecology, genetics, neonatology and, if available, social work, nursing and medical ethics.

The American Academy of Pediatrics Committee on Genetics. (2000) Molecular Genetic Testing in Pediatric Practice: A Subject Review. *Pediatrics.* 106: 1494-1497.
This statement on molecular genetic testing in pediatric practice reviews various types of genetic testing that may be encountered in pediatric practice and provides guidelines for the pediatrician. Due to the complex nature of genetic testing and pretest counseling

about the indications, benefits and limits of testing, the AAP considers expert interpretation and explanation of genetic testing results to individuals and families as essential. The AAP cautions that presymptomatic testing or carrier screening, particularly for diseases with serious health implications, can have profound effects and should not be performed without pretest counseling. According to the AAP, "central to all types of genetic testing is the process of genetic counseling to ensure that the patient has adequate information to give truly informed consent, that he or she is psychologically prepared to cope with the results, and that patients and sometimes other family members receive assistance in understanding the medical, psychological, social, and legal implications of these findings."

Hersh, JH, and The American Academy of Pediatrics Committee on Genetics. (2008) Health Supervision for Children With Neurofibromatosis. *Pediatrics.* 121: 633-642.
This set of guidelines, published by the AAP, is designed to assist pediatricians in caring for children with Neurofibromatosis Type 1. The AAP recommends genetic counseling in the neonatal/prenatal period. Pediatricians should discuss the genetics of NF1 or refer the adolescent for genetic counseling between the ages of 13-21. When there is uncertainty regarding a definitive diagnosis, consideration should be given to seeking genetic consultation and determining whether genetic testing is indicated at that time to expedite a diagnosis.

The American Academy of Pediatrics Committee on Genetics. (1999) Folic Acid for the Prevention of Neural Tube Defects. *Pediatrics.* 104: 325-327.
This practice guideline discusses the use of folic acid for the prevention of NTDs. The AAP indicates that high-risk patients should be cautioned that folic acid supplementation does not preclude the need for counseling or consideration of prenatal testing for NTDs.

The American Academy of Pediatrics Committee on Genetics. (2002) Health Supervision for Children with Sickle Cell Disease. *Pediatrics.* 109: 526-525.
This set of guidelines, published by the AAP, is designed to assist pediatricians in caring for children with sickle cell disease. The AAP recommends the provision of genetic counseling for individuals with sickle cell disease during middle childhood and adolescence. They note that, "it is important that education and genetic counseling be provided by professionals with expertise in genetics and in the clinical manifestations and treatment of SCD." They also indicate that referral to a hematologist-oncologist or a clinical geneticist or obstetrician associated with a prenatal diagnosis may be appropriate for the discussion of prenatal diagnosis.

Plauche´ Johnson, C , Myers, CM, and The American Academy of Pediatrics Council on Children With Disabilities. (2007) Identification and Evaluation of Children with Autism Spectrum Disorders. *Pediatrics.* 120: 1183-1215.
This AAP recommendation is intended to assist clinicians with the identification and evaluation of autism spectrum disorders. The AAP indicates that genetic counseling regarding recurrence risk in siblings of children with ASD is important even when the etiologic evaluation is negative.

The American Academy of Pediatrics Committee on Bioethics. (2001) Ethical Issues with Genetic Testing in Pediatrics. *Pediatrics.* 107: 1451-1455.
In their discussion of ethical issues surrounding pediatric genetic testing, the AAP indicates that the complexity of the issues surrounding genetic testing indicates the need for detailed counseling, informed consent, and confidentiality as part of the genetic testing process. They also recommend detailed genetic counseling for predictive testing for late-onset disorders.

The American Academy of Pediatrics Committee on Genetics. (2008) Maternal Phenylketonuria. *Pediatrics.* 122: 445-449.
All individuals, particularly women and girls of childbearing age, should be referred to an experienced PKU treatment center for genetic and nutritional evaluation and counseling throughout their lifetime. Genetic counseling should be offered for all women with PKU before and after conception.

Kaye, CI and The American Academy of Pediatrics Committee on Genetics. (2006) Newborn Screening Fact Sheets. *Pediatrics.* 118: e934-e963.
The newborn screening fact sheets reviews several common conditions included in newborn screening programs. The AAP indicates that management of all persons identified with congenital hearing loss requires a comprehensive genetic evaluation and that core personnel include individuals with expertise in the genetics of hearing loss, dysmorphology, audiology, otolaryngology, and genetic counseling. They also recommend that parents of children identified with CF through newborn screening require education on all aspects of CF and that genetic counseling should be arranged for all such families. Their discussion of homocystinuria indicates that genetic counseling and screening should be offered to relatives of persons with homocystinuria.

Trotter, TL, Hall, JG and The American Academy of Pediatrics Committee on Genetics. (2005) Health Supervision for Children With Achondroplasia. *Pediatrics.* 116: 771-783.
This set of guidelines, published by the AAP, is designed to assist pediatricians in caring for children with Achondroplasia and indicates the need for provision of genetic counseling throughout the duration of the life of a child with achondroplasia.

2. American Congress of Obstetricians and Gynecology

The American Congress of Obstetricians and Gynecologists. (2007) Practice Bulletin No. 78: Hemoglobinopathies in Pregnancy. *Obstetrics & Gynecology*. 109: 229-238.
ACOG reviews the most common hemoglobinopathies and provides recommendations for screening and clinical management of hemoglobinopathies during pregnancy. ACOG indicates that individuals of African, Southeast Asian and Mediterranean descent should be offered carrier screening for hemoglobinopathies. Genetic counseling is recommended if both parents are determined to be carriers. These couples that are at risk for a having a child with a hemoglobinopathy may benefit from genetic counseling to review the natural history of these disorders, prospects for treatment and cure, their risk, availability of prenatal genetic testing and reproductive options.

The American Congress of Obstetricians and Gynecologists. (2007) Practice Bulletin No. 77: Screening for Fetal Chromosomal Abnormalities. *Obstetrics & Gynecology*. 109: 217-228.
ACOG presents information regarding the use of ultrasonographic and serum markers for selected aneuploidy screening in pregnancy and provides practical recommendations for implementing Down syndrome screening in practice. ACOG recommends that: "Women found to have increased risk of aneuploidy with first-trimester screening should be offered genetic counseling and the option of CVS or second trimester amniocentesis. An abnormal finding on second-trimester ultrasound examination identifying a major congenital anomaly significantly increases the risk of aneuploidy and warrants further counseling and the offer of a diagnostic procedure."

The American Congress of Obstetricians and Gynecologists. (2006) Committee Opinion No. 338: Screening for Fragile X Syndrome. *Obstetrics & Gynecology*. 107: 1483-1485.
ACOG presents a review of Fragile X syndrome and provides practical recommendations for Fragile X syndrome screening in practice. ACOG recommends that patients with a family history of mental retardation or a history of fragile X mental retardation should receive genetic counseling and should be offered genetic testing to assess their risk for having an affected child. Additionally genetic counseling should be considered for a women with an intermediate number of triplet repeats identified through molecular genetic testing or discordancy between the triplet repeat number and the methylation status.

The American Congress of Obstetricians and Gynecologists. (2008) Committee Opinion No. 409: Direct-to-Consumer Marketing of Genetic Testing. *Obstetrics & Gynecology*. 111: 1493-1494.
In their discussion of direct-to-consumer marketing of genetic testing, ACOG indicates that all genetic testing should be provided only after consultation with a qualified health care professional, and that for complex testing, this may involve referral to a genetic counselor or a medical geneticist. Appropriate pretest and posttest counseling should be provided, including a discussion of the risks, benefits and limitations of the testing.

The American Congress of Obstetricians and Gynecologists. (2008) Committee Opinion No. 410: Ethical Issues in Genetic Testing. *Obstetrics & Gynecology*. 111: 1495-1502.
ACOG reviews some of the ethical issues related to genetic testing and provides guidelines for the appropriate use of genetic tests by obstetrician–gynecologists. ACOG indicates that patients found to have genetic susceptibility to cancer should be offered counseling and follow-up. ACOG also asserts that: "Obstetrician–gynecologists should recognize that geneticists and genetic counselors are an important part of the health care team and should consult with them and refer as needed."

The American Congress of Obstetricians and Gynecologists. (2009) Committee Opinion No. 446: Array Comparative Genomic Hybridization in Prenatal Diagnosis. *Obstetrics & Gynecology*. 114: 1161-1163.
ACOG provides a discussion of and guidelines for the use of array CGH in prenatal diagnosis. Targeted array CGH, in concert with genetic counseling, can be offered in prenatal cases with abnormal anatomic findings and a normal conventional karyotype, as well as in cases of fetal demise with congenital anomalies and the inability to obtain a conventional karyotype. ACOG indicates that couples choosing targeted array CGH should receive both pretest and posttest genetic counseling and that follow-up genetic counseling is required for interpretation of array CGH results.

The American Congress of Obstetricians and Gynecologists. (2009) Committee Opinion No. 442: Preconception and Prenatal Carrier Screening for Genetic Diseases in Individuals of Eastern European Jewish Descent. *Obstetrics & Gynecology*. 114: 950-953.
ACOG for and provides revised recommendations for carrier screening for individuals of Eastern European Jewish descent and clinical management of identified carriers. In addition, ACOG addresses the other carrier screening tests that are available for this population and management of screening for such conditions. It is recommended that four core conditions be offered to individuals in this population. ACOG indicates that individuals of Eastern European Jewish descent should be offered carrier screening for Cystic Fibrosis, Tay Sachs, Canavan Disease and Familial Dysautonomia, and that genetic counseling is recommended if both parents are determined to be carriers. Genetic counseling can assist such couples by reviewing the natural history of these disorders, prospects for treatment and cure, their risk, availability of prenatal genetic testing and reproductive options. There is carrier testing available for additional conditions that are more prevalent in this population. ACOG indicates that patient education materials can be made available to assist in informed decision about having additional screening tests and that some patients may benefit from genetic counseling. Individuals with a positive family history of one of these disorders should be offered carrier screening for the specific disorder and may benefit from genetic counseling. When both partners are carriers of one of these disorders, they should be referred for genetic counseling and offered prenatal diagnosis. In addition, patients having difficulty with the decision of whether to pursue additional carrier screening may be assisted in making an informed

decision about carrier testing through counseling by a genetic counselor, geneticist, or physician with expertise in these diseases.

The American Congress of Obstetricians and Gynecologists. (2009) Committee Opinion No. 432: Spinal Muscular Atrophy. *Obstetrics & Gynecology*. 113: 1194-1196.
While ACOG's Committee on Genetics does not recommend preconception and prenatal screening for SMA in the general population at this time, they review the indications under which testing should be offered and discuss appropriate clinical management of such cases. The Congress recommends that genetic counseling for SMA carrier screening be offered to couples with a family history SMA or SMA-like disease and to those couples who request SMA carrier screening. In addition, all identified carriers for SMA should be referred for follow-up genetic counseling for a discussion of risk to the fetus and future pregnancies. Patients requesting fetal testing for SMA should be referred to an appropriate provider of prenatal genetic counseling and testing services and if needed, referral for medical and genetic counseling should be made for patients with a fetus found to be affected with SMA.

The American Congress of Obstetricians and Gynecologists. (2009) Practice Bulletin No. 103: Hereditary Breast and Ovarian Cancer Syndrome. *Obstetrics & Gynecology*. 113: 957-966.
The ACOG practice bulletin on hereditary breast and ovarian cancer provides guidelines that can assist an obstetrician–gynecologist in determining which individuals might benefit from a more thorough hereditary cancer risk assessment. The Congress indicates that hereditary cancer risk assessment is conducted by a health care provider with expertise in cancer genetic. The Congress indicates situations that may warrant referral for genetic counseling, including an isolated case of breast cancer at or before age 50 years in a patient with limited family structure. The Congress indicates that a genetic risk assessment is recommended for patients with a greater than an approximate 20–25% chance of having an inherited predisposition to breast cancer and ovarian cancer.

The American Congress of Obstetricians and Gynecologists. (2009) Practice Bulletin No. 102: Management of Stillbirth. *Obstetrics & Gynecology*. 113: 748-761.
This ACOG bulletin reviews the current information on stillbirth. The Congress recommends genetic counseling in the event of a stillbirth if follow-up studies indicate the presence of a family genetic condition.

The American Congress of Obstetricians and Gynecologists. (2006) Committee Opinion No. 350: Breast Concerns in the Adolescent. *Obstetrics & Gynecology*. 108: 1329-1336.
This ACOG committee opinion on breast concerns in the adolescent addresses a variety of breast issues that pertain to the adolescent patient and provides guidance to clinicians in managing such patients. The Congress indicates that adolescent patients with a family history of breast cancer that are interested in genetic testing should be referred for genetic counseling and that counseling by an appropriately qualified individual is advised for patients considering genetic testing.

The American Congress of Obstetricians and Gynecologists. (2005) Committee Opinion #325: Update on Carrier Screening for Cystic Fibrosis. *Obstetrics & Gynecology*. 106: 1465-1468.
This ACOG committee opinion discusses the updated cystic fibrosis carrier screening recommendations. These recommendations discuss a variety of situations where genetic counseling is beneficial, including: if a woman is a carrier of a cystic fibrosis mutation and her partner is unavailable for testing, couples with the R117H mutation, for individuals with a family history of cystic fibrosis, when both partners are cystic fibrosis carriers, carrier testing that identifies individuals with two cystic fibrosis mutations who have not previously received a diagnosis of cystic fibrosis.

The American Congress of Obstetricians and Gynecologists. (2005) Committee Opinion #318: Screening for Tay-Sachs Disease. *Obstetrics & Gynecology*. 106: 893-894.
ACOG provides a review of Tay Sachs Disease and recommendations for carrier screening for Tay Sachs disease. ACOG indicates that individuals of Ashkenazi Jewish, French–Canadian, or Cajun descent and those with a family history consistent with TSD should be offered carrier screening for TSD. Genetic counseling should be offered if both parents are determined to be carriers. These couples that are at risk for a having a child with TSD may benefit from genetic counseling to review the natural history of this disorder, prospects for treatment and cure, their risk, availability of prenatal genetic testing and reproductive options.

The American Congress of Obstetricians and Gynecologists. (2005) Perinatal Risks Associated With Assisted Reproductive Technology. *Obstetrics & Gynecology*. 106: 1143-1146.
This ACOG committee opinion reviews perinatal risks associated with ART. The Committee recommends that all patients with congenital bilateral absence or atrophy of the vas deferens and their partners considering IVF by sperm extraction procedure with ICSI should be offered genetic counseling to discuss testing for cystic fibrosis, due to the association with these features and this genetic condition.

3. American College of Medical Genetics

Toriello, HV, for the Professional Practice and Guidelines Committee of the American College of Medical Genetics. (2005) Folic Acid and Neural Tube Defects. *Genetics in Medicine.* 7: 283-284.
ACMG Practice Guideline provides a review of neural tube defects and folic acid and offers recommendations to guide care. ACMG recommends that: "Women who have had a prior NTD-affected pregnancy, who have a first-degree relative with a NTD, or who are themselves affected should obtain genetic counseling concerning their occurrence or recurrence risks, pregnancy management and the appropriate folic acid intake for them."

The American College of Medical Genetics. Statement on Universal Newborn Hearing Screening. January 2000. Available on the World Wide Web at: http://www.acmg.net/resources/policies/pol-023.asp. Accessed September 28, 2009.
This ACMG statement on universal newborn hearing screening indicates that it is, "essential that all children with confirmed hearing loss be referred for evaluation and genetic counseling to a team which will typically include a qualified clinical geneticist and a genetic counselor."

MAKE THESE WEB REFERENCES LIKE THE ONES IN SECTION 1

The American College of Medical Genetics. Statement on Direct-to-Consumer Genetic Testing. April 2008. Available on the World Wide Web at: http://www.acmg.net/AM/Template.cfm?Section=Policy_Statements&Template=/CM/HTMLDisplay.cfm&ContentID=4157. Accessed September 28, 2009.
This ACMG statement discusses direct-to-consumer genetic testing and some of the concerns that the organizations has with these services. The College indicates that minimally, genetic testing should involve a knowledgeable professional, for the process of ordering and interpreting a genetic test. The College states that genetics expert such as a certified medical geneticist or genetic counselor "can help the consumer avoid a number of risks including lack of informed consent, inappropriate testing, misinterpretation of results, testing that is inaccurate or not clinically valid, lack of follow-up care, misinformation, and other adverse consequences."

The American College of Medical Genetics Subcommittee on Cystic Fibrosis Screening. (2001) Laboratory Standards and Guidelines for Population Based Cystic Fibrosis Carrier Screening. *Genetics in Medicine.* 3: 149-154.
This ACMG statement provides guidance for the clinician with regards to cystic fibrosis carrier screening. The College indicates that various cystic fibrosis test results will generate the need for genetic counseling, including, "the identification of positive/negative couples who may request additional mutation analyses or counseling to clarify their residual risk, individuals who have a family history of CF, otherwise healthy males who carry mutations or variants associated with infertility, an positive/positive couples." Because of the subtle and complicated genetic issues raised, the Committee recommends that the detection of the R117H and 5T polymorphism be followed by

referral of the patient to a geneticist or other expert professional for further counseling and appropriate testing. For patients with a positive family history and a negative population based carrier screening or with an admixture of various ethnic groups, the Committee indicates that accurate risk assessment requires genetic counseling. The College also recommends that any primary care provider who does not feel comfortable explaining concepts associated with cystic fibrosis carrier screening to the patients should refer them to a genetics professional.

The American College of Medical Genetics Professional Practice and Guidelines Committee. (2008) Carrier Screening in Individuals of Ashkenazi Jewish Descent. *Genetics in Medicine*. 10: 54-56.
This ACMG statement provides guidance for the clinician with regards to carrier screening for individuals of Ashkenazi Jewish descent. The College indicates that formal genetic counseling and medical genetic consultation should be readily available to any patient in this population desiring this service. In the case where someone is identified as a carrier, genetic counseling should be readily available to discuss the findings and possible reproductive options. Furthermore, a discussion regarding the importance of genetic counseling for other family members should be stressed.

The American Society of Human Genetics and The American College of Medical Genetics. (1995) Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents. *American Journal of Human Genetics.* 57: 1233 -1241.
This ACMG statement provides a discussion of the risks, benefits, and limitations of genetic testing in children and adolescents. The College indicates that: "Educations and counseling for parents and the child, commensurate on maturity, should precede genetic testing," and that, "follow-up genetic counseling should be readily available."

The American College of Medical Genetics Work Group on Management of Pompe Disease. (2006) Pompe Disease Diagnosis and Management Guidelines. *Genetics in Medicine*. 8:267-288.
This ACMG practice guideline discusses the diagnosis and management of Pompe disease. The College indicates that: "Pompe disease is a multi-system disorder and is best managed by a multidisciplinary team that includes a genetic counselor." In addition, the College recommends that genetic counseling should be offered to all parents with an affected child with Pompe disease and to all adults with Pompe disease.

The American College of Medical Genetics and American Society of Human Genetics. (2000) Genetic Testing for Colon Cancer. *Genetics in Medicine.* 2: 362-366.
This joint statement provides an overview of hereditary colon cancer syndromes and genetic testing and provides recommendations for appropriate genetic testing protocol for at risk patients. The ACMG and ASHG indicate that genetic counseling is an important and crucial component of the genetic risk assessment process, and that a clear understanding by the patient can only be arrived at by careful counseling.

Byers PH, Krakow D, Nunes ME, Pepin M. (2006) Genetic Evaluation of Suspected Osteogenesis Imperfecta (OI). *Genetics in Medicine.* 8: 383-388.
This statement by the ACMG provides a recommended approach to the diagnosis of OI. The College indicates that genetic evaluation and counseling is recommended for confirmation and discussion about natural history, treatment and prenatal diagnosis in future pregnancies.

Prior TW. For the American College of Medical Genetics Professional Practice Committee. (2008) Carrier Screening for Spinal Muscular Atrophy. *Genetics in Medicine.* **10: 840-842.**
The ACMG provides a review of SMA, genetic testing for the detection of SMA carriers, and recommendations for clinicians in managing carrier screening for patients. The College recommends that SMA carrier testing be offered to all couples regardless of race or ethnicity and that formal genetic counseling services must be made available to anyone requesting this testing. In addition, all identified carriers should be referred for follow-up genetic counseling for a discussion of risk to the fetus or future pregnancies.

Driscoll DA, for the American College of Medical Genetics Professional Practice and Guidelines Committee. (2004) Second Trimester Maternal Serum Screening for Fetal Open Neural Tube Defects and Aneuploidy. *Genetics in Medicine.* 6: 540-541.
The ACMG reviewed the current recommendations for second trimester maternal serum screening for open neural tube defects and aneuploidy. The College recommends that genetic counseling and/or educational material should be available to patients to review the different screening tests that are available, and that women should be counseled regarding the advantages and limitations of the available tests. The College indicates that genetic counseling and additional testing such as targeted ultrasound examination and amniocentesis are recommended for pregnancies with an elevated MSAFP test result and for patients who are screen positive for either trisomy 21 or 18.